Arizona Department of Health Services Newborn Screening Program Report Table Changes

TSH: Abnormal or Borderline

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	TSH = 84.7 μU/mL	Abnormal – See Comments	< 30 μU/mL
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FA	Normal Findings	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT within Normal Limits	Normal Findings	See Reverse

CAH: Abnormal or Borderline

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	17-OHP = 57.6 ng/mL	Borderline Abnormal – See Comments	See Reverse
Hemoglobinopathies	FA	Normal Findings	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT within Normal Limits	Normal Findings	See Reverse

Hemoglobinopathies:

Disease

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FS	See Comments (Hemoglobin)	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT within Normal Limits	Normal Findings	See Reverse

Traits

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FAS	See Comments (Hemoglobin)	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT within Normal Limits	Normal Findings	See Reverse

Biotinidase: Pale or Abnormal

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FA	Normal Findings	FA
Biotinidase Deficiency	Slight enzyme activity present	See Comments	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT within Normal Limits	Normal Findings	See Reverse

Galactosemia: Abnormal or Equivocal

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FA	Normal Findings	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Equivocal	Equivocal – See Comments	Full Enzyme Activity
Amino Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT within Normal Limits	Normal Findings	See Reverse

MSMS Disorders (Amino Acid Disorders, Fatty Acid Oxidation Disorders, & Organic Acid Disorders)

Pilot project disorders: Abnormal

MSUD (LEU), Homocystinuria (MET), C3, IVA (C5), & GA-1 (C5DC)

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FA	Normal Findings	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	LEU = 320 µmol/L VAL = 450 µmol/L LEU/PHE = 5.00 VAL/PHE = 6.00	See Comments (MSUD)	< 280.00 μmol/L < 340.00 μmol/L < 4.70 < 5.20
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT within Normal Limits	Normal Findings	See Reverse

Elevated primary analyte not indicative of disorder

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FA	Normal Findings	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	MET = 200 μmol/L MET/PHE = 1.00	See Comments (Elevated)	< 75.00 μmol/L < 1.40
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT within Normal Limits	Normal Findings	See Reverse

Interfering Agent not indicative of disorder

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FA	Normal Findings	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	PHE = 200 μmol/L MET = 200 μmol/L MET/PHE = 1.00	See Comments (Interfering Agent)	< 181.80 μmol/L < 75.00 μmol/L < 1.40
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT within Normal Limits	Normal Findings	See Reverse

Non-Pilot project disorders: Abnormal

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FA	Normal Findings	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Fatty Acid Oxidation Disorders	C8 = 0.87 μmol/L	See Comments (MCAD)	< 0.60 μmol/L
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT within Normal Limits	Normal Findings	See Reverse

Cystic Fibrosis:

Elevated IRT, no DNA mutations

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FA	Normal Findings	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT = 70.2 ng/mL DNA = no mutations	Elevated IRT No mutations detected	< 65 ng/mL No Mutations detected

HIGH IRT, no DNA mutations

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FA	Normal Findings	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT = 150.2 ng/mL DNA = no mutations	High IRT >= 150 ng/mL No mutations detected	< 65 ng/mL No Mutations detected

Heterozygous, Double Heterozygous, and Homozygous mutation(s)

Disorder Group	Result Value	Interpretation	Reference Range
Congenital Hypothyroidism (CH)	Within Normal Limits	Normal Findings	See Reverse
Congenital Adrenal Hyperplasia (CAH)	Within Normal Limits	Normal Findings	See Reverse
Hemoglobinopathies	FA	Normal Findings	FA
Biotinidase Deficiency	Within Normal Limits	Normal Findings	Full Enzyme Activity
Galactosemia	Within Normal Limits	Normal Findings	Full Enzyme Activity
Amino Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Fatty Acid Oxidation Disorders	Within Normal Limits	Normal Findings	See Reverse
Organic Acid Disorders	Within Normal Limits	Normal Findings	See Reverse
Cystic Fibrosis	IRT = 93.7 ng/mL DNA= D1270N	One mutation detected	< 65 ng/mL No Mutations detected